16 Inheritance and variation

No two living organisms are identical, they all show **variation**. Much of this variation is passed on from one generation to the next via **genes**.

An introduction to chromosomes and genes

Chromosomes

Chromosomes are present in the nuclei of all living cells. Each chromosome is composed of a single **deoxyribonucleic acid (DNA) molecule** wrapped around **proteins** called **histones**. DNA molecules contain **genetic information** in the form of **genes**. In any cell that is not dividing, chromosomes exist as long, thin strands known as **chromatin threads** which are spread throughout the nucleus. Chromosomes become visible when a cell begins to divide due to them becoming shorter and thicker.

Chromosomes are passed on from one generation to the next in **gametes** and each species has a distinctive **number** of chromosomes per body cell, for example, every human cell has 46 chromosomes. The number of chromosomes in each cell is known as the **diploid number** or 2***n*** **number**. Chromosomes exist in pairs known as **homologous pairs**. Every human cell has 23 pairs, one member of each pair being of **maternal origin** and the other of **paternal origin**. With the exception of the pair of sex chromosomes, members of each pair look alike.

Genes

Genes are specific sections of chromosomal DNA molecules and are the basic units of **hereditary**. Each human body cell has over 30,000 genes and each gene controls a particular characteristic. Genes work by controlling the production of **protein** in cells, mainly the production of **enzymes**. Each gene controls the production of a specific protein.

All the cells of one organism contain an **identical combination** of genes. It is this combination that makes each organism **unique** since no two organisms, except identical twins or organisms produced asexually from one parent, have the same combination of genes. Within any cell some genes are active while others are inactive, e.g. in a nerve cell, genes controlling the activity of the nerve cell are active and genes that would control the activity of a muscle cell are inactive.

Cell division

When a cell divides, **chromosomes** with their **genes** are passed on to the cells produced, known as **daughter cells**. There are two types of cell division, **mitosis** and **meiosis**.

Mitosis

Mitosis occurs in **all body cells** except in the formation of gametes. During mitosis, **two genetically identical** cells are formed. Each cell contains the **same** number of chromosomes as the parent cell, i.e. the **diploid number**.

Mitosis is important because:

- It ensures that each daughter cell contains the **diploid** number of chromosomes. This maintains the **species number** of chromosomes in all members of a species.
- It ensures that each daughter cell has an **identical** combination of genes.
- It is the method by which all cells of a multicellular organism are formed, hence it is essential for **growth** and to **repair** damaged tissues.
- It is the method by which organisms reproduce **asexually** forming offspring that are **identical** to each other and to the parent.

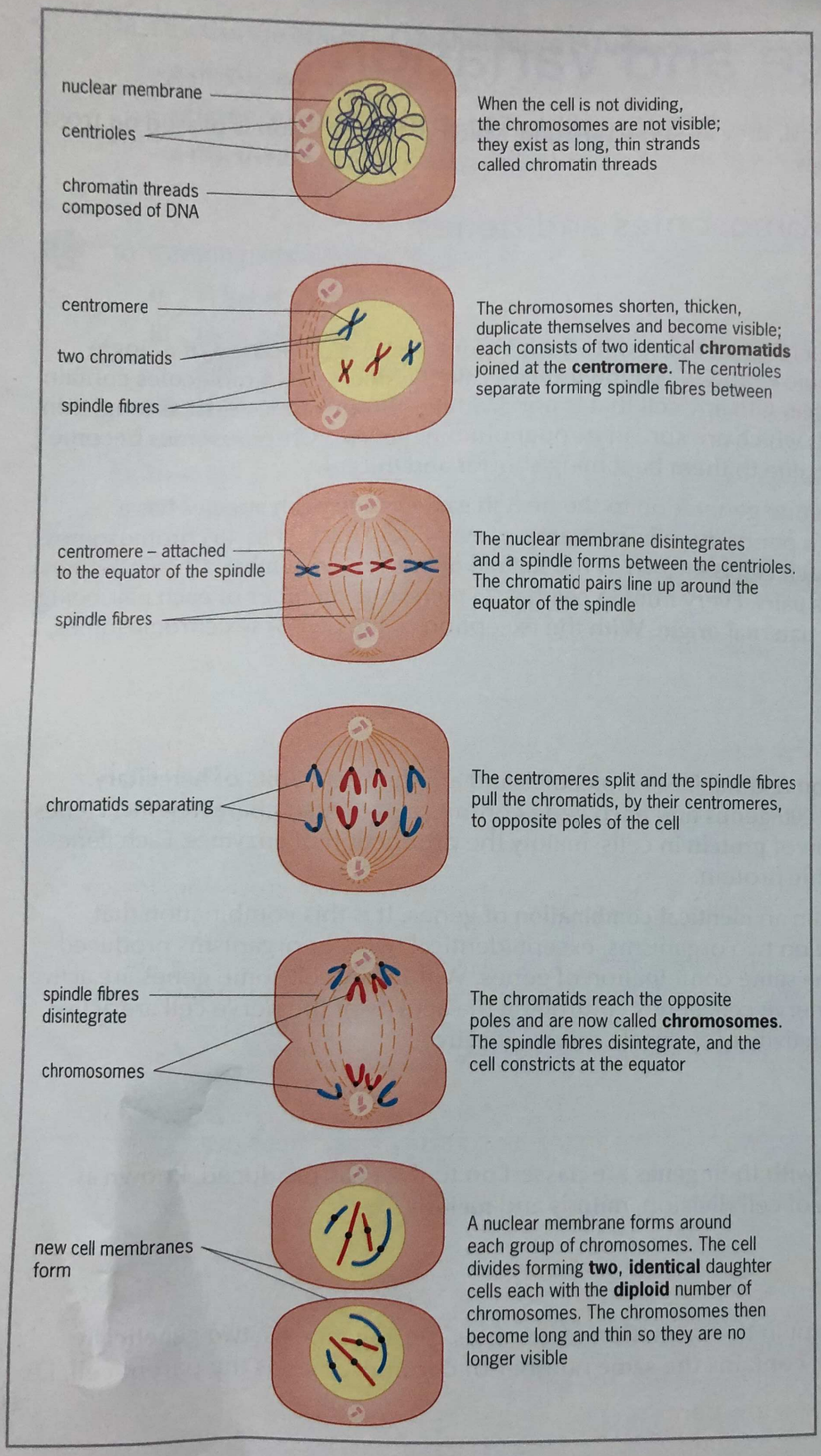


Figure 16.1 *The process of mitosis in an animal cell with four chromosomes; two of paternal origin (blue) and two of maternal origin (red)*

Mitosis and asexual reproduction in plants

Some plants can reproduce **asexually** by **mitosis** occurring in certain structures of the parent plant, a process known as **vegetative propagation**. Since mitosis produces genetically identical cells, all offspring produced asexually from one parent are **genetically identical** and are collectively called a **clone**. **Cloning** is the process of making genetically identical organisms through non-sexual means.

Examples of natural vegetative propagation

- New plants can grow from **vegetative organs** at the beginning of the growing season, e.g. from rhizomes, stem tubers, corms and bulbs (see page 89).
- New plants can grow from **outgrowths** of the parent plant, e.g. from runners, leaf buds and suckers.

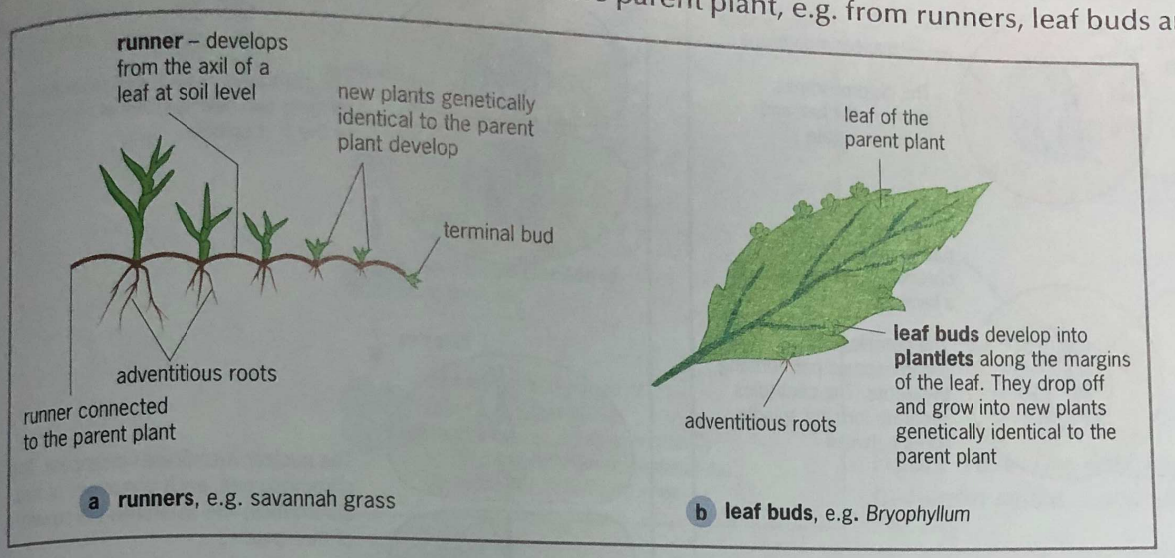


Figure 16.2 *Examples of vegetative propagation in plants*

Examples of artificial vegetative propagation

- By taking **cuttings**, farmers and gardeners artificially propagate plants. Cuttings are parts of plants that will develop roots and shoots to become new plants if given suitable conditions, e.g. **stem cuttings**.
  - When a piece of a **sugar cane** stem with two or three buds is placed horizontally on the soil, new plants grow from each bud.
  - When a stem of **hibiscus** plant with a few leaves at the top is planted, roots grow from the cut end forming a new plant.
- **Tissue culture** is used to artificially propagate plants, e.g. to propagate orchids, potatoes and tomatoes. Small pieces of tissue called **explants** are taken from a parent plant and grown in a nutrient-rich culture, under sterile conditions, to form cell masses known as **calluses**. Each callus is then stimulated with appropriate plant hormones to grow into a new plant.

If cuttings or explants are taken from plants with **desirable characteristics**, e.g. a high yield, high quality, resistance to disease or fast growth rate, then all plants produced will have the same desirable characteristics.

Cloning in animals

To **clone** an animal, a nucleus is removed from an ovum of a female donor. A cell, still containing its nucleus, is taken from the animal to be cloned and is fused with the ovum. This newly created ovum is placed into a surrogate mother where it is stimulated to develop into an embryo. The surrogate then gives birth to a new individual that is **genetically identical** to the animal from which the original cell came, e.g. Dolly the sheep. A very low percentage of cloned embryos survive to birth, and animals born alive often have health problems or other abnormalities, and reduced life spans.

Meiosis (reduction division)

Meiosis occurs only in the **reproductive organs** during the production of **gametes**. During meiosis, **four genetically non-identical** cells are formed. Each cell contains **half** the number of chromosomes as the parent cell, known as the **haploid number** or ***n* number**.

Meiosis is important because it ensures that:

- Each daughter cell has the **haploid** number of chromosomes. The **diploid** number can then be restored at **fertilisation**.

- Each daughter cell has a **different** combination of genes. This leads to **variation** among offspring which enables species to constantly change and adapt to changing environmental conditions (see page 156).

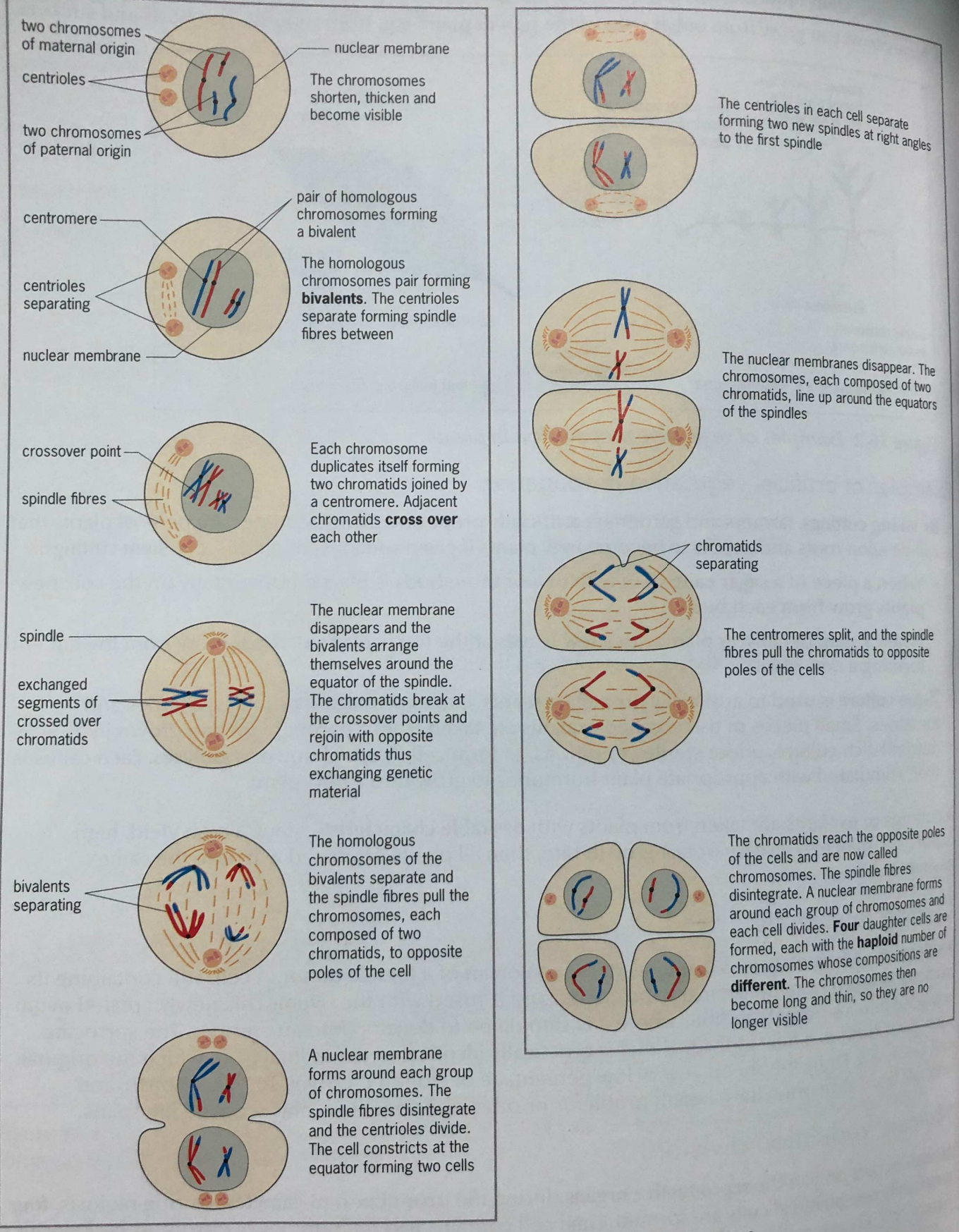


Figure 16.3 *The process of meiosis in an animal cell with four chromosomes*

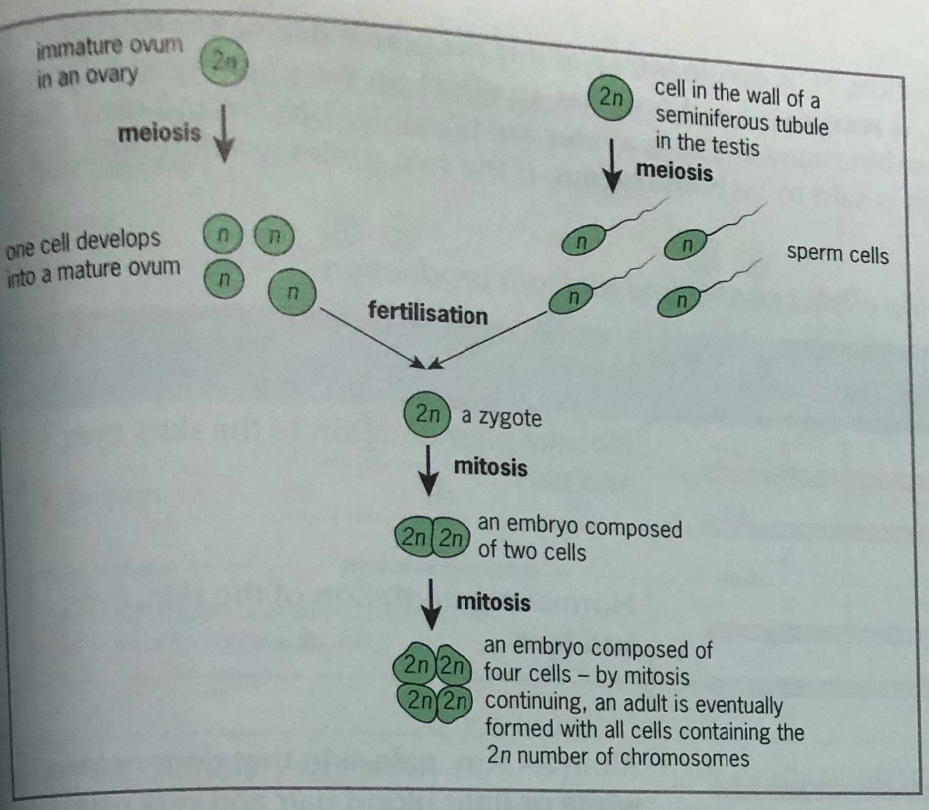


Figure 16.4 *The relationship between mitosis and meiosis*

Revision questions

1. Outline the relationship between chromosomes and genes.
2. Outline the process of mitosis in an animal cell.
3. Give THREE reasons why mitosis is important to living organisms.
4. Describe TWO different natural ways that plants can reproduce asexually.
5. Where does meiosis occur in living organisms?
6. In what ways does meiosis differ from mitosis?
7. Give TWO reasons why meiosis is important to living organisms.

Inheritance

Like chromosomes, genes exist in **pairs**. One gene of each pair is of **maternal origin** and one is of **paternal origin**, and the pairs occupy equivalent positions on homologous chromosomes. A gene controlling a particular characteristic can have different forms known as **alleles**. Each gene usually has two different alleles.

The **composition of genes** within the cells of an organism makes up the organism's **genotype**. The **observable characteristics** of an organism make up its **phenotype**.

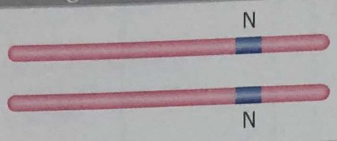
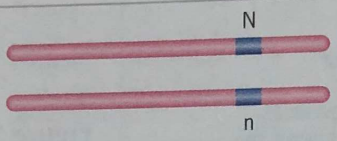
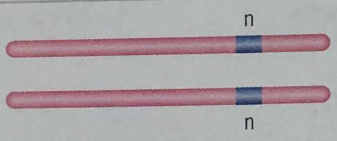
Example: albinism in humans

People with **albinism** produce very little or no melanin in their skin, eyes and hair. The gene controlling the production of the pigment melanin has **two** different **alleles** which can be represented using letters:

- N stimulates melanin production
- n fails to stimulate melanin production

The allele stimulating melanin production, **N**, is **dominant**, i.e. if it is present it shows its effect on the phenotype. The allele for albinism, **n**, is **recessive**, i.e. it only has an effect on the phenotype if there is no dominant allele present. Three combinations of these alleles are possible; **NN**, **Nn** and **nn**. If the two alleles are the same, the organism is said to be **homozygous**. If the two alleles are different, the organism is said to be **heterozygous**.

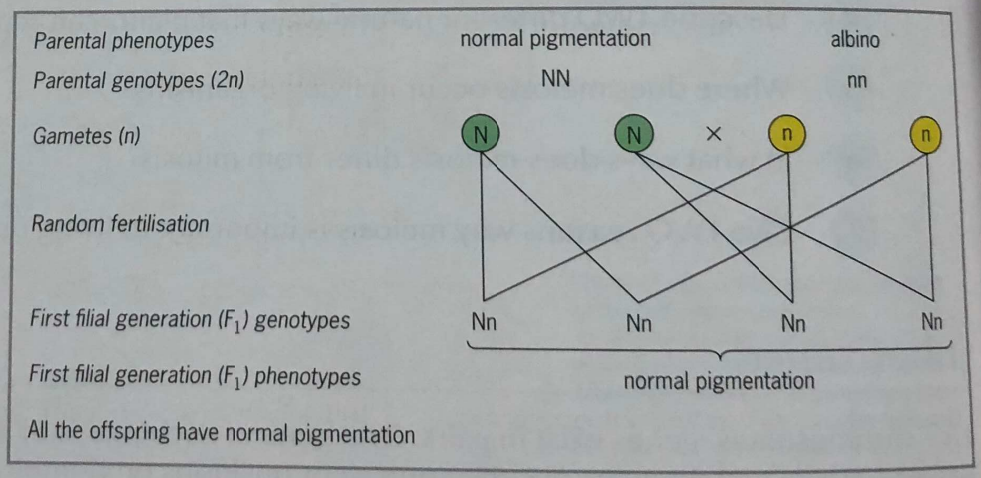
Table 16.1 *Possible combinations of the alleles controlling melanin production*

Genotype (combination of alleles)	How the alleles appear on homologous chromosomes	Phenotype (appearance)
NN **Homozygous dominant** (pure breeding)		Normal pigmentation of the skin, eyes and hair
Nn **Heterozygous** (carrier)		Normal pigmentation of the skin, eyes and hair
nn **Homozygous recessive** (pure breeding)		Albino – very pale skin that does not tan, white or light blond hair and very pale blue eyes

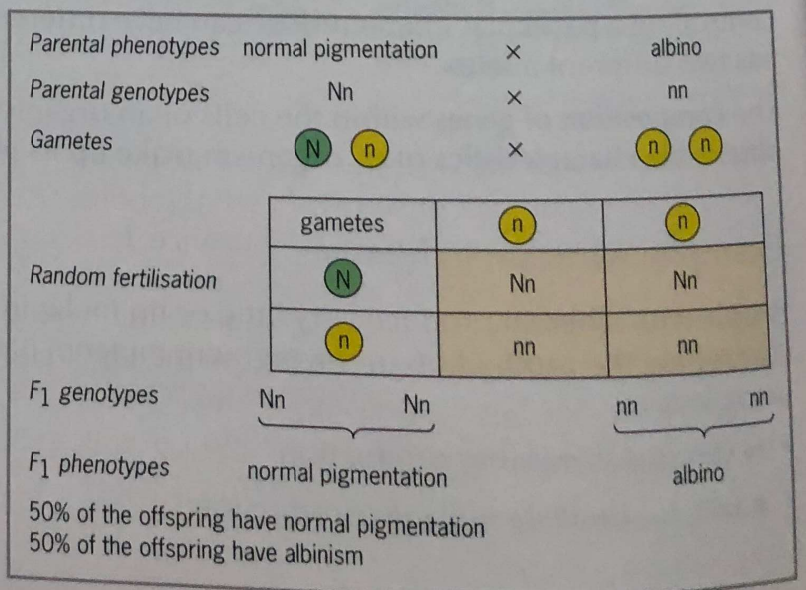
Gametes produced in meiosis contain only **one chromosome** from each homologous pair. As a result, they contain only **one allele** from each pair. When fertilisation occurs, chromosomes and the alleles they carry **recombine** to form pairs in the zygote.

Results of possible crosses

1 If one parent is **homozygous dominant** and one is **homozygous recessive**:



2 If one parent is **heterozygous** and one is **homozygous recessive**, showing the use of a **Punnett square** to predict the outcome of the cross:



3 If both parents are **heterozygous**, i.e. carriers:

Parental phenotypes	normal pigmentation	×	normal pigmentation
Parental genotypes	Nn	×	Nn
Gametes	N n	×	N n

Random fertilisation

gametes	N	n
N	NN	Nn
n	Nn	nn

F_1 genotypes: NN, Nn, Nn, nn

F_1 phenotypes: normal pigmentation, normal pigmentation, normal pigmentation, albino

75% of the offspring have normal pigmentation
25% of the offspring have albinism

Table 16.2 *Summary of genotypic and phenotypic ratios of offspring from different crosses*

Genotype of parents	Genotypic ratio of offspring	Phenotypic ratio of offspring
homozygous dominant × homozygous dominant	100% homozygous dominant	**all** show the **dominant** trait
homozygous dominant × heterozygous	50% homozygous dominant, 50% heterozygous	**all** show the **dominant** trait
homozygous dominant × homozygous recessive	100% heterozygous	**all** show the **dominant** trait
heterozygous × heterozygous	25% homozygous dominant, 50% heterozygous, 25% homozygous recessive	**75%** show the **dominant** trait, **25%** show the **recessive** trait i.e. a **3:1** ratio
heterozygous × homozygous recessive	50% heterozygous, 50% homozygous recessive	**50%** show the **dominant** trait, **50%** show the **recessive** trait i.e. a **1:1** ratio
homozygous recessive × homozygous recessive	100% homozygous recessive	**all** show the **recessive** trait

Co-dominance

Sometimes neither allele dominates the other such that the influence of both alleles is visible in the heterozygous individual. These alleles show **co-dominance**. For example, in the impatiens plant, allele **R** stimulates the production of **red** flowers and allele **W** stimulates the production of **white** flowers. When a plant with **red** flowers, genotype **RR**, is crossed with one with **white** flowers, genotype **WW**, all the F_1 generation have **pink** flowers with the genotype **RW**. Other examples include:

- **sickle cell anaemia**
- **ABO blood groups.**

Impatiens plants with pink flowers

Sickle cell anaemia

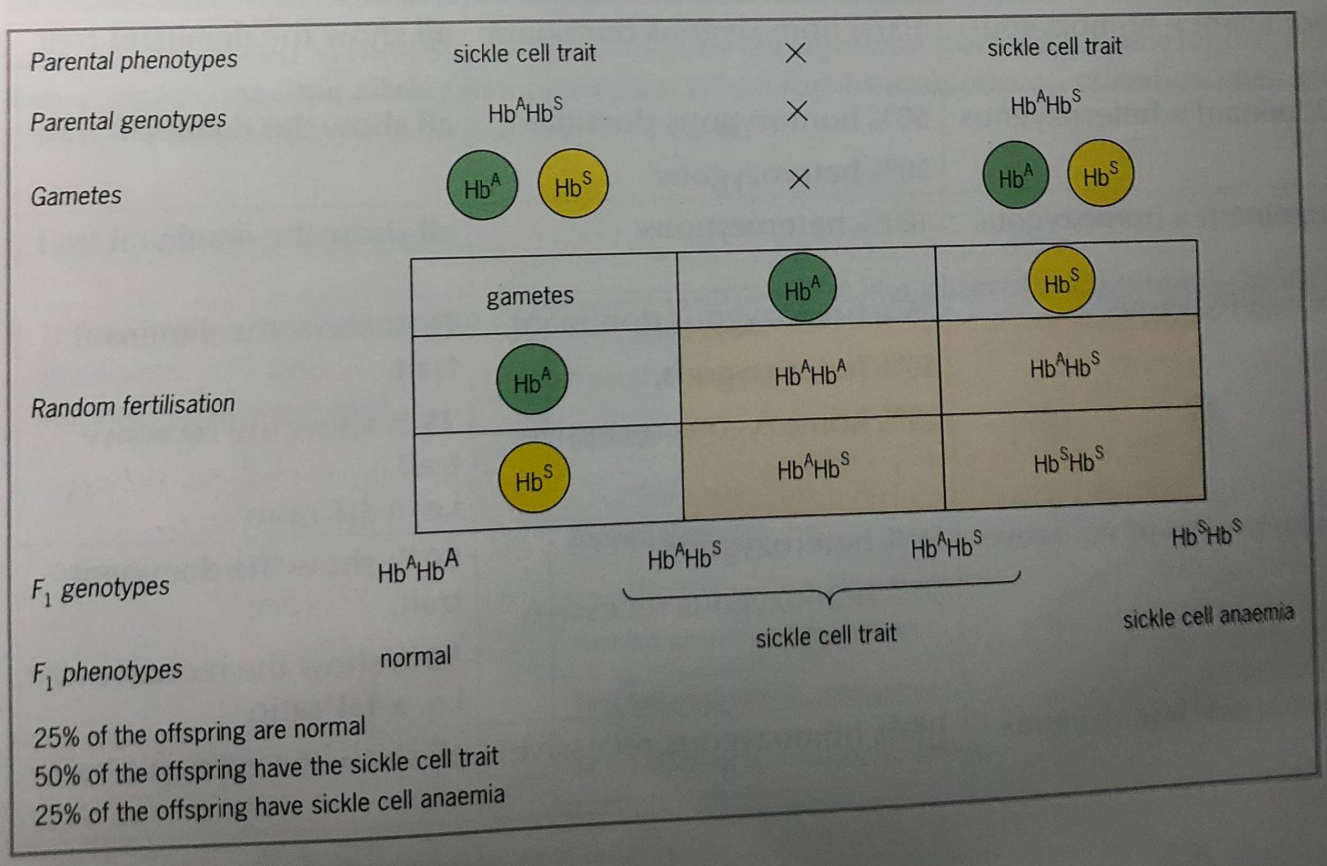
The blood of a person with sickle cell anaemia contains abnormal **haemoglobin S** instead of normal **haemoglobin A**. The disease is caused by an abnormal allele. The normal allele Hb^A stimulates the production of normal haemoglobin A, the abnormal allele Hb^S stimulates the production of abnormal haemoglobin S. These alleles show co-dominance.

Table 16.3 *Possible combinations of alleles controlling haemoglobin production*

Genotype	Haemoglobin produced	Phenotype
Hb^A Hb^A	100% haemoglobin A	**Normal.**
Hb^A Hb^S	55–65% haemoglobin A 35–45% haemoglobin S	**Sickle cell trait.** Usually no symptoms. Symptoms of sickle cell anaemia may develop in very low oxygen concentrations, e.g. at high altitude or during extreme physical exercise.
Hb^S Hb^S	100% haemoglobin S	**Sickle cell anaemia.** Symptoms of sickle cell anaemia develop which include painful crises, anaemia, increased vulnerability to infections and jaundice.

Example

If both parents have **sickle cell trait:**



ABO blood groups

ABO blood groups are controlled by **three** alleles, I^A, I^B and I^O.

- I^A and I^B are both dominant to I^O.
- I^A and I^B are co-dominant, i.e. there is no dominance between them.

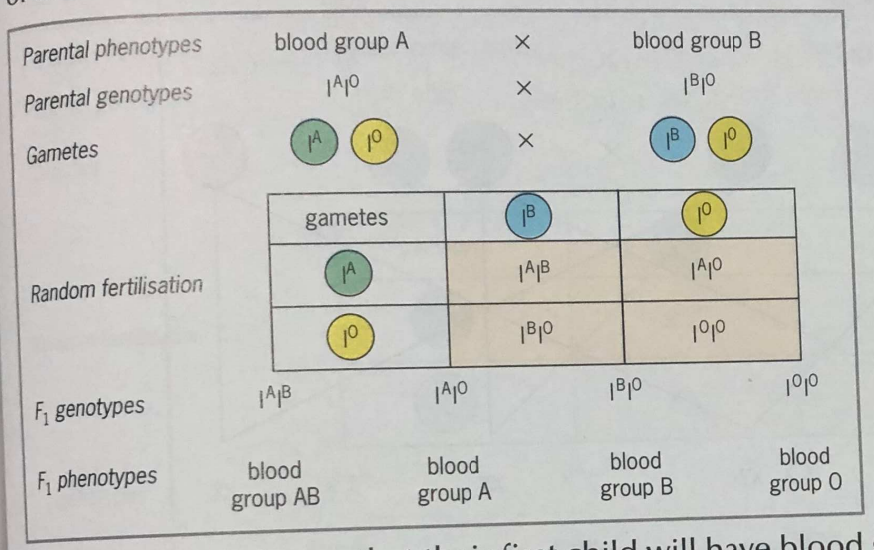
Only **two** alleles are present in any cell.

Table 16.4 *Possible combinations of alleles controlling ABO blood groups*

Genotype	Phenotype
$I^A I^A$	Blood group A
$I^A I^O$	Blood group A
$I^B I^B$	Blood group B
$I^B I^O$	Blood group B
$I^A I^B$	Blood group AB
$I^O I^O$	Blood group O

Sample question

A **heterozygous** female of **blood group A** marries a **heterozygous** male of **blood group B**. What are the chances of their first child having blood group O? Explain your answer by means of a genetic-cross diagram.



There is a **1 in 4** chance that their first child will have blood group O.

Pedigree charts

A **pedigree chart** shows how a specific trait is passed down among family members. Pedigree charts can be used to determine **genotypes**, or possible genotypes, of the individuals shown, and to predict possible genotypes and phenotypes of future offspring. This information is used by **genetic counsellors** to identify potential risks for future offspring developing a genetic disorder.

Example

A chemical substance called **PTC** tastes bitter to some people and is tasteless to others. The ability to taste PTC is controlled by a pair of alleles. The allele enabling tasting of PTC, **T**, is dominant. The non-tasting allele, **t**, is recessive.

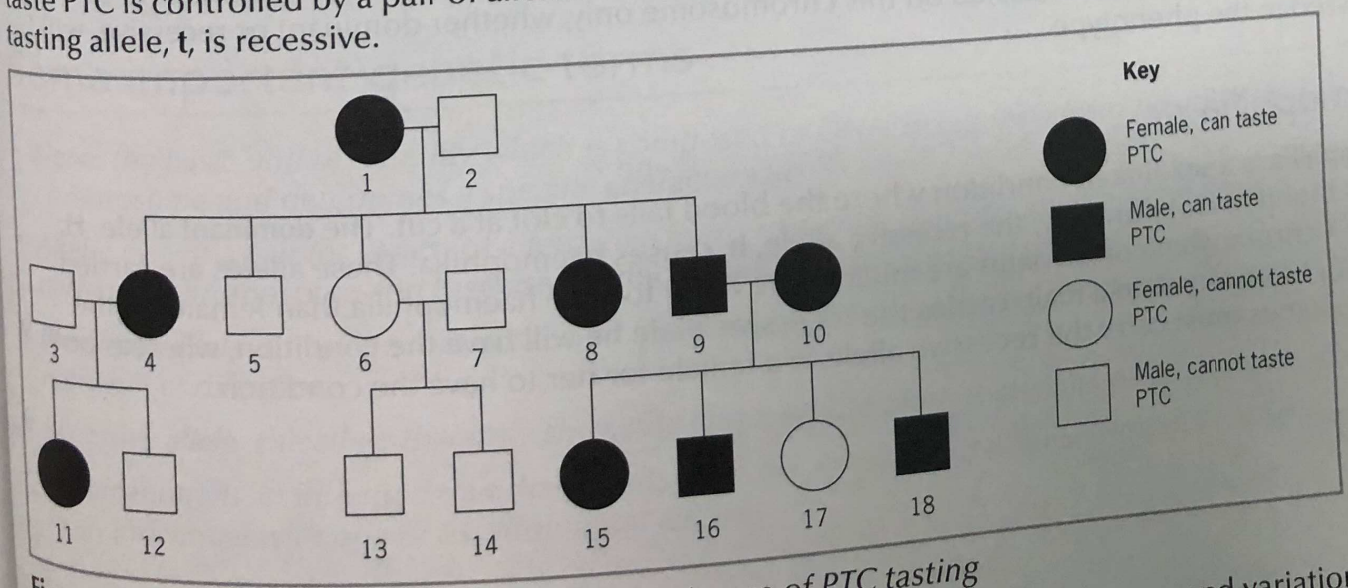
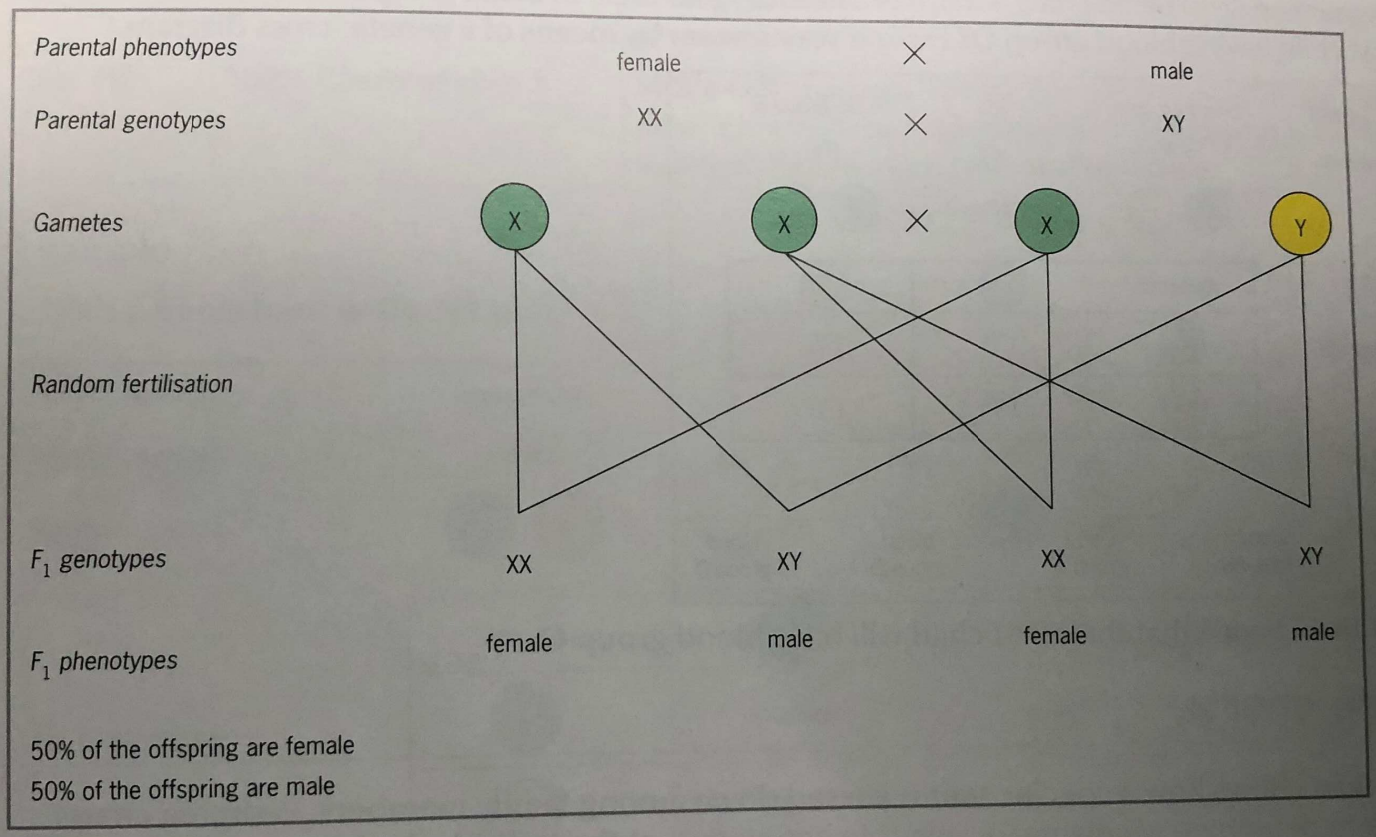


Figure 16.5 *A pedigree chart to show the inheritance of PTC tasting*

- Genotypes of individuals 2, 3, 5, 6, 7, 12, 13, 14, and 17 must be **tt** since they all have the recessive trait.
- Genotypes of individuals 1, 4, 8, 9, 10 and 11 must be **Tt** since they all had one parent who had the recessive trait or produced at least one offspring with the recessive trait.
- Genotypes of individuals 15, 16 and 18 could be **TT** or **Tt** since both parents were heterozygous.

Mechanism of sex determination

In each cell, one pair of chromosomes is composed of the **sex chromosomes**. There are two types, **X** and **Y**, and they determine the individual's gender. Genotype **XX** is **female**; genotype **XY** is **male**. Only the **male** can pass on the **Y** chromosome, consequently the **father** is the parent who determines the gender of his offspring.



Sex-linked characteristics

Sex-linked characteristics are characteristics determined by genes carried on the sex chromosomes that have nothing to do with determining gender. These are known as **sex-linked genes**. Since chromosome X is longer than chromosome Y, it carries more genes. Males only have one X chromosome and any allele carried on this chromosome only, whether dominant or recessive, will b expressed in the phenotype.

Haemophilia

Haemophilia is a sex-linked condition where the blood fails to clot at a cut. The **dominant** allele, **H**, causes blood to clot normally; the **recessive** allele, **h**, causes haemophilia. These alleles are carried on the **X** chromosome only. Males are much more likely to have haemophilia than females; if the single X chromosome in a male carries the recessive allele he will have the condition, whereas both chromosomes must carry the recessive allele in a female for her to have the condition.

Table 16.5 *Possible combinations of alleles controlling blood clotting*

Genotype	Phenotype
$X^H X^H$	Female, normal blood clotting
$X^H X^h$	Female, normal blood clotting (carrier)
$X^h X^h$	Female with haemophilia
$X^H Y$	Male, normal blood clotting
$X^h Y$	Male with haemophilia

Example

A cross between a **female** with **normal blood clotting** who is a **carrier**, and a **male** with **normal blood clotting**.

Parental phenotypes	female, normal clotting	×	male, normal clotting
Parental genotypes	$X^H X^h$	×	$X^H Y$
Gametes	X^H X^h	×	X^H Y

Random fertilisation

gametes	X^H	Y
X^H	$X^H X^H$	$X^H Y$
X^h	$X^H X^h$	$X^h Y$

F_1 genotypes	$X^H X^H$	$X^H Y$	$X^H X^h$	$X^h Y$
F_1 phenotypes	female, normal clotting	male, normal clotting	female, normal clotting	male with haemophilia

All the female offspring have normal blood clotting
50% of the male offspring have normal blood clotting
50% of the male offspring have haemophilia

Colour blindness

Colour blindness is a sex-linked condition where the sufferer is unable to distinguish differences between certain colours. The **dominant** allele, **N**, allows normal vision and the **recessive** allele, **n**, causes colour blindness. These alleles are carried on the **X** chromosome only, so colour blindness is inherited in the same way as haemophilia.

Some important genetic terms

- **Gene:** the basic unit of heredity which is composed of DNA, occupies a fixed position on a chromosome and determines a specific characteristic.
- **Allele:** one of a pair (or series) of alternative forms of a gene that occupy the same position on a particular chromosome and that control the same characteristic.
- **Dominant allele:** the allele that, if present, produces the same phenotype whether its paired allele is identical or different.
- **Recessive allele:** the allele that only shows its effect on the phenotype if its paired allele is identical.
- **Dominant trait:** an inherited trait that results from the presence of a single dominant allele. It is seen in an individual with one or two dominant alleles.

- ***Recessive trait:*** *an inherited trait that results from the presence of two recessive alleles. It is only seen in an individual with no dominant allele.*
- ***Co-dominance:*** *neither allele dominates the other such that the influence of both alleles is visible in the heterozygous individual.*
- ***Genotype:*** *the combination of alleles present in an organism.*
- ***Phenotype:*** *the observable characteristics of an organism.*
- ***Homozygous:*** *having two identical alleles in corresponding positions on a pair of homologous chromosomes.*
- ***Heterozygous:*** *having two different alleles in corresponding positions on a pair of homologous chromosomes.*

Variation

No two living organisms are exactly alike, not even identical twins. **Variation** arises from a combination of **genetic causes** and **environmental causes**. The **phenotype** of an organism is determined by its **genotype** and the influences of its **environment**:

phenotype = genotype + environmental influences

Genetic causes of variation

Genetic variation arises in several ways:

- **Meiosis**. Every gamete produced by meiosis has a different combination of genes as a result of:
  - chromatids of homologous chromosomes crossing over and **exchanging** genes
  - chromosomes arranging themselves around the equators of the spindles in totally **random** ways.
- **Sexual reproduction.** During **fertilisation**, male and female gametes fuse in completely **random ways** to create different combinations of genes in each zygote.
- **Mutations.** A mutation is a sudden **change** in a single gene or in part of a chromosome containing several genes. Mutations cause new characteristics to suddenly develop in organisms. Mutations occurring in body cells cannot be inherited whereas mutations occurring in a gamete or zygote can be inherited. Most mutations are harmful; however, a few produce **beneficial characteristics** which provide the organism with a **selective advantage** in the struggle for survival, e.g. the peppered moth (see page 159).

Environmental causes of variation

Living organisms are constantly affected by the different factors in their **environment.** Food, drugs, physical forces, temperature and light can affect animals. Temperature, light intensity, availability of mineral salts and water all affect plants. This variation is not caused by genes and **cannot** be passed on to offspring.

The importance of variation

Variation is important because:

- It enables species to **adapt** to changing environmental conditions, improving their chances of survival.
- It provides the raw material on which **natural selection** can work, and is therefore essential for species to remain **well adapted** to their environment or to gradually **change** and **improve** by becoming better adapted to their environment.
- It makes it **less likely** that any adverse changes in environmental conditions will wipe out an entire species since some organisms may be able to adapt to the new conditions.

Types of variation

There are two basic types of variation within a species:

- **Continuous variation**

Continuous variation is where characteristics show a **continuous gradation** from one extreme to the other without a break. Most organisms fall in the middle of the range with fewer at the two extremes, i.e. the characteristics show a **normal distribution**. Examples include height, weight, foot size, hair colour, and leaf size in plants.

Characteristics showing continuous variation are usually controlled by **many genes** and can be affected by environmental factors.

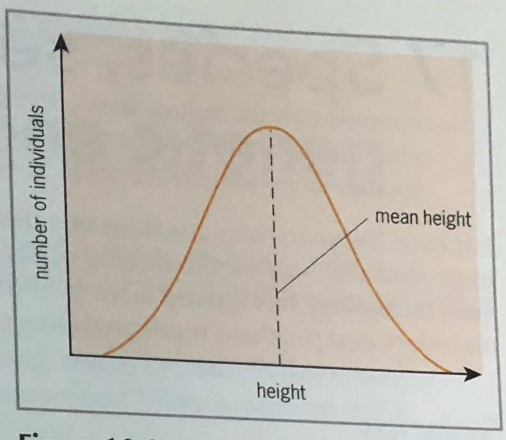


Figure 16.6 *A normal distribution curve showing height*

- **Discontinuous variation**

Discontinuous variation is where characteristics show **clear cut differences** with no intermediates. Individuals can be divided into distinct categories, e.g. ABO blood groups, tongue rolling, and the presence or absence of horns in cattle. Characteristics showing discontinuous variation are usually controlled by a **single gene** and environmental factors have little, if any, influence on them.

Revision questions

8 Distinguish between the following pairs of terms:

a gene and allele

b genotype and phenotype

c homozygous and heterozygous.

9 PTC is a chemical that tastes bitter to some people and is tasteless to others. The ability to taste PTC is dominant. Use appropriate symbols and a genetic-cross diagram to show how a couple who can both taste PTC can produce a child who is unable to taste PTC.

10 What is co-dominance?

11 Is it possible for a female of blood group A and a male of blood group AB to have a child of blood group B? Use appropriate symbols and a genetic-cross diagram to support your answer.

12 In humans, is it the mother or father who determines the sex of their children? Explain your answer by means of a genetic diagram.

13 What are sex-linked characteristics?

14 Colour blindness is caused by an X-linked, recessive allele. Two parents with normal colour vision have a colour blind child. Use a genetic diagram to show how this is possible (X^N = normal vision; X^n = colour blindness).

15 Outline THREE ways in which genetic variation arises.

16 Give THREE reasons why it is important that living organisms show variation.

17 By reference to specific examples, distinguish between continuous and discontinuous variation.